# Copy number variant analysis of human embryonic stem cells.

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**Public Summary:**

**Scientific Abstract:**
Differences between individual DNA sequences provide the basis for human genetic variability. Forms of genetic variation include single-nucleotide polymorphisms, insertions/duplications, deletions, and inversions/translocations. The genome of human embryonic stem cells (hESCs) has been characterized mainly by karyotyping and comparative genomic hybridization (CGH), techniques whose relatively low resolution at 2-10 megabases (Mb) cannot accurately determine most copy number variability, which is estimated to involve 10%-20% of the genome. In this brief technical study, we examined HSF1 and HSF6 hESCs using array-comparative genomic hybridization (aCGH) to determine copy number variants (CNVs) as a higher-resolution method for characterizing hESCs. Our approach used five samples for each hESC line and showed four consistent CNVs for HSF1 and five consistent CNVs for HSF6. These consistent CNVs included amplifications and deletions that ranged in size from 20 kilobases to 1.48 megabases, involved seven different chromosomes, were both shared and unique between hESCs, and were maintained during neuronal stem/progenitor cell differentiation or drug selection. Thirty HSF1 and 40 HSF6 less consistently scored but still highly significant candidate CNVs were also identified. Overall, aCGH provides a promising approach for uniquely identifying hESCs and their derivatives and highlights a potential genomic source for distinct differentiation and functional potentials that lower-resolution karyotype and CGH techniques could miss. Disclosure of potential conflicts of interest is found at the end of this article.

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